



SYNGAP1 gene

synaptic Ras GTPase activating protein 1

Normal Function

The *SYNGAP1* gene provides instructions for making a protein, called SynGAP, that plays an important role in nerve cells in the brain. SynGAP is found at the junctions between nerve cells (synapses) where cell-to-cell communication takes place. Connected nerve cells compose the "wiring" in the circuitry of the brain. Synapses are able to change and adapt over time, rewiring brain circuits, which is critical for learning and memory. SynGAP helps regulate synapse adaptations and promotes proper brain wiring. The protein's function is particularly important during a critical period of early brain development that affects future cognitive ability.

Health Conditions Related to Genetic Changes

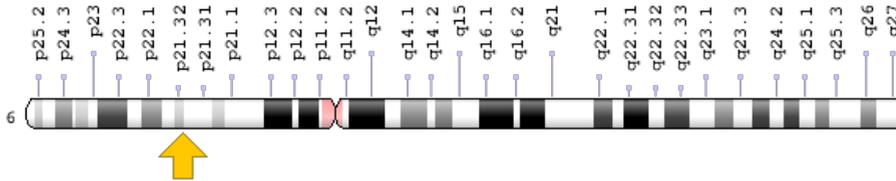
SYNGAP1-related intellectual disability

At least 40 mutations in the *SYNGAP1* gene have been found to cause *SYNGAP1*-related intellectual disability. In addition to mild-to-moderate intellectual disability, this condition commonly features other neurological problems, including recurrent seizures (epilepsy) and developmental disorders that affect communication and social interaction (autism spectrum disorders). Gene mutations involved in *SYNGAP1*-related intellectual disability prevent the production of functional SynGAP protein from one copy of the gene, reducing the protein's activity in cells. Studies show that a reduction of SynGAP activity can have multiple effects in nerve cells, including pushing synapses to develop (mature) too early. The changes triggered by a reduction of SynGAP activity disrupt the synaptic adaptations in the brain that underlie learning and memory, leading to cognitive impairment and other neurological problems characteristic of *SYNGAP1*-related intellectual disability.

Chromosomal Location

Cytogenetic Location: 6p21.32, which is the short (p) arm of chromosome 6 at position 21.32

Molecular Location: base pairs 33,420,070 to 33,453,689 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIAA1938
- MRD5
- neuronal RasGAP
- Ras GTPase-activating protein SynGAP
- ras/Rap GTPase-activating protein SynGAP
- RASA5
- synaptic Ras GTPase-activating protein 1
- synaptic Ras GTPase activating protein 1 homolog
- synaptic Ras GTPase activating protein, 135kDa
- SYNGAP

Additional Information & Resources

Educational Resources

- Basic Neurochemistry: Molecular, Cellular and Medical Aspects (sixth edition, 1999): Synaptic Plasticity as a Model for Learning and Memory Research <https://www.ncbi.nlm.nih.gov/books/NBK27983/>
- Madame Curie Bioscience Database (2000): Introduction to Branching Morphogenesis in Vertebrate Neurons https://www.ncbi.nlm.nih.gov/books/NBK6520/#_A46741_

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SYNGAP1%5BTIAB%5D%29+OR+%28synaptic+Ras+GTPase+activating+protein+1%5BTIAB%5D%29%29+OR+%28%28Ras+GTPase-activating+protein+SynGAP%5BTIAB%5D%29+OR+%28SYNGAP%5BTIAB%5D%29+OR+%28neuronal+RasGAP%5BTIAB%5D%29+OR+%28ras/Rap+GTPase-activating+protein+SynGAP%5BTIAB%5D%29+OR+%28synaptic+Ras+GTPase+activating+protein+1+homolog%5BTIAB%5D%29+OR+%28synaptic+Ras+GTPase+activating+protein,+135kDa%5BTIAB%5D%29+OR+%28synaptic+Ras+GTPase-activating+protein+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SYNAPTIC RAS-GTPase-ACTIVATING PROTEIN 1
<http://omim.org/entry/603384>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SYNGAP1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SYNGAP1%5Bgene%5D>
- HGNC Gene Family: C2 and RasGAP domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/830>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11497
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8831>
- UniProt
<http://www.uniprot.org/uniprot/Q96PV0>

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Reviewed: May 2016

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services